Choanal Atresia, Posterior

Alternative Names
PCA

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities > Congenital malformations of the respiratory system

Incidence per 100,000 Live Births
11-50

OMIM Number
608911

Mode of Inheritance
Probably a multifactorial trait, autosomal recessive inheritance also reported

Gene Map Locus
N/A

Description
Choanal atresia is a rare developmental anomaly characterized by failure of communication of the posterior nasal cavity with the nasopharynx. It may be complete or incomplete, unilateral (60%) or bilateral (40%), bony (90%) or membranous (10%) or a combination of these.

The anomaly presents either immediately after birth as respiratory distress, or as a coincidental finding at an older age. The condition is the most common nasal abnormality, affecting in 1 out of every 7,000 to 8,000 live births, with a 2:1 female-to-male ratio.

Molecular Genetics
N/A

Epidemiology in the Arab World

Egypt
Sadek (1998) reported two females from a consanguineous family from Egypt with bilateral congenital choanal atresia. The patients also presented features of vitamin D resistant rickets. Their mother had beta-thalassemia minor.

Al-Gazali et al. (2003) reported a male infant from unrelated Egyptian parents, residents of the United Arab Emirates, with Raine syndrome. There were no histories of any congenital anomaly or genetic disease or of maternal drug intake during pregnancy. Prenatal ultrasound showed polyhydramnios and short limbs. The baby presented at birth with severe craniofacial anomalies including a wide anterior fontanel, exophthalmos, severe depression of the nasal bridge with a hypoplastic midface, bilateral choanal atresia, and a large protruding tongue.

Kuwait
Petkovska et al. (2007) identified seven children (five girls), including one of a monozygotic twin pair, with congenital choanal atresia during a 3-year period (2001-2004). Five of these patients had bilateral choanal atresia, while in two it was unilateral. It was membranous and osseous in two patients each, and of mixed type in the remaining three. Two of the patients had an associated cleft palate, and one of these two had a right lung agenesis. Of the seven, two fit the criteria for CHARGE association.

Palestine
Gershoni-Baruch (1992) described a small inbred Muslim kindred from Palestine in which nonsyndromal choanal atresia occurred in two sibs and their paternal uncle. In a note added in proof, she reported the birth of a third affected sib.

Al-Gazali et al. (2002) reported a brother and sister from an inbred Palestinian family with an autosomal recessive syndrome of choanal atresia, hypothyelia/athelia, and thyroid gland anomalies. The parents were first cousins and their first couple was a female with bilateral choanal atresia who died few hours after birth. There was a history of two miscarriages at two months of gestation. The parents and four children were normal. The male patient had, at birth, a unilateral right choanal
atresia, low set ears, hypoplastic nipples, and absent breast tissue. The affected sister cried immediately after birth but three minutes later collapsed and needed incubation and resuscitation. She had widely spaced eyes with depressed nasal bridge, dysmorphic low set ear, a small pit on the upper right side of the neck, sparse eyebrows, and long eyelashes. Both nipples were hypoplastic and there was no breast tissue. She had bilateral choanal atresia. Chromosome study was normal. She died at 4 months of age following a pneumonia. Al-Gazali et al. (2002) noted that the features overlapped those of Bambforth syndrome, hypohidrotic ectodermal dysplasia with hypothyroidism and ciliary dyskinesia (HEDH) syndrome, and methimazole embryopathy.

Lebanon
Topley et al (1995) described a Lebanese infant with choanal atresia who was born in the UAE for non-consanguineous parents. The data were collected from Tawam hospital where the infant received the surgical repair. Also, the parents were questioned for a history of illness or drug consumption during pregnancy, and for a family history of congenital abnormalities and consanguinity. Choanal atresia was thought to be the consequence of the interference with the migration of neural crest cells, and the influence of chemical and genetic factors. [See also: United Arab Emirates > Topley et al., 1995].

Oman
Verma and Siddiqui (2002) reported bilateral choanal atresia in a 15-year old female who presented with longstanding nasal obstruction since childhood. Endoscopic examination of the nose revealed thick mucoid secretions at the floor of both nasal cavities, with complete obstruction of the lumen posteriorly, and no communication between the nose and nasopharynx, which was confirmed by CT scan. Endonasal correction of bilateral choanal atresia was performed using CO2 laser for soft tissues and nasal micro-drill for the bony atresia. No stent was required, as the lumen was adequately wide. The patient was discharged the next day with the advice of regular douching of the nasal cavity. Upon follow up after a year, there was a wide posterior choana with no stenosis or recurrence of the atresia.

Saudi Arabia
Rejjal et al. (1994) studied CT scans of the brain in 23 consecutive patients with congenital choanal atresia (CCA; 10 boys and 13 girls) to determine the prevalence and the scope of associated brain abnormalities. CCA was associated with known syndromes or different congenital abnormalities in 7 and 10 patients, respectively, and was an isolated finding in 6 patients. Six patients (26%) had brain abnormalities including alobar holoprosencephaly, absent corpus callosum, benign enlargement of the subarachnoid space of infancy, cerebral calcification, small right hemisphere, small ventricles and microcephaly. None of the brain abnormalities were suspected clinically. Brain abnormalities were more common in boys compared to girls (40% vs 15.4%, p = 0.18), in bilateral compared to unilateral CCA (31.3% vs 14%, p = 0.38), and in membranous compared to bony CCA (75% vs 15.7%, p = 0.02).

Syria
Sadek (1998) reported a Syrian male with bilateral congenital choanal atresia. The patient also presented features of congenital inguinal hernia and bilateral pre-auricular sinuses. His parents were second cousins and he had one brother with bilateral congenital choanal atresia. Two more brothers and three sisters were all normal.

United Arab Emirates
Topley et al (1995) identified three infants with choanal atresia and one infant with choanal stenosis who were born during a 5-month period at Tawam hospital in Al Ain. Thus, file examination of the infants with choanal atresia in the past six years was performed at the same hospital. Two other national infants were detected to have choanal atresia in the past six years who were not born in Tawam hospital. Also, parents were questioned for a history of illness or drug consumption during pregnancy, and for a family history of congenital abnormalities and consanguinity. The diagnosis was based on the failure to pass a naso-gastric tube through the nostril followed by contrast nasography. In addition, some patients presented respiratory difficulties. All patients were receiving the surgical repair in Tawam hospital with age ranged from four days to six years. In most cases, delayed repair was due to the need for treatment of associated anomalies. One case of the clustered group had an Indian mother, and two cases had consanguineous parents. All mothers of the clustered group took some medication during pregnancy. Two of them took oral contraceptive, one mother took iron and an antacid, and the fourth mother took paracetamol, a cephalosporin, meclocizine, and pyridoxine. In the clustered group, one patient had Down syndrome, and two had CHARGE association (the combination of choanal atresia, cardiovascular, urogenital, neurodevelopmental, and craniofacial anomalies). Choanal atresia and the associated anomalies were thought to be the consequences of the interference with the migration of neural crest cells, and the influence of chemical and genetic factors. Topley et al (1995) were unable to explain the presence of
the cluster group with choanal atresia, however, viral and other environmental teratogen cannot be excluded.

Sadek (1998) reported a male from the United Arab Emirates with bilateral congenital choanal atresia. The patient was born to first cousin parents who were clinically normal. His mother had five spontaneous abortions at 12 weeks, two babies who died at 5 months of age from congenital hydronephrosis, and two still births at 8 months gestation with multiple congenital anomalies. He also had a sister with unilateral choanal atresia.

Nawaz et al. (1998) reviewed the experience in the United Arab Emirates in the management of esophageal atresia (EA) and tracheoesophageal fistula (TEF). In their review, Nawaz et al. (1998) studied 41 patients with EA and/or TEF. Approximately 51% of the patients had associated congenital malformations, including choanal atresia.

[See also: Palestine > Al-Gazali et al., 2002].

Yemen
In an inbred Yemenite family, Qazi et al. (1982) observed posterior choanal atresia in a brother and sister and their paternal aunt. All four parents of the three affected persons traced to a common ancestral couple 2-3 generations earlier. In 1998, Sadek reported two males and one female from a non-consanguineous Yemeni family with bilateral congenital choanal atresia. The patients and their mother all had features of Crouzon syndrome. There were three normal siblings in the family (two brothers and one sister).

References

Related CTGA Records
Down Syndrome
Hemoglobin--Beta Locus
Holoprosencephaly
Osteosclerotic Bone Dysplasia, Lethal
Tracheoesophageal Fistula with or without Esophageal Atresia

External Links
http://www.childsdoc.org/fall98/choanal/choanal.asp

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